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Teaching Neuro*Images*: Coats disease revealing facioscapulohumeral muscular dystrophy

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A 21-year-old Brazilian woman presented with mild facial muscle weakness, scapular winging and an asymmetric shoulder girdle, and peroneal weakness. Medical history disclosed mildly symptomatic Coats disease (CD) (figure). Muscle MRI studies were highly suggestive of facioscapulohumeral muscular dystrophy type 1 (FSHD1) (figure) and genetic testing confirmed this diagnosis.

FSHD1 represents an autosomal dominant muscular dystrophy with typical muscle MRI patterns associated with extramuscular manifestations, including sensorineural deafness and complex retinal vasculopathy with telangiectasis, such as CD.[1] CD may be a presenting feature of FSHD1 with high risk of exudative retinal detachment, retinal microaneurysm formation, and peripheral retinal telangiectasis.[2]

AUTHOR CONTRIBUTIONS

P.V.S. Souza: case report project: conception, organization, execution; manuscript: writing of the first draft. W.B.V.R. Pinto: case report project: conception, organization, execution; manuscript: writing of the first draft. G.N.R. Batistella: case report project: execution; manuscript: writing of the first draft. R.B. Dias: case report project: execution; manuscript: writing of the first draft. A.S.B. Oliveira: manuscript: review and critique.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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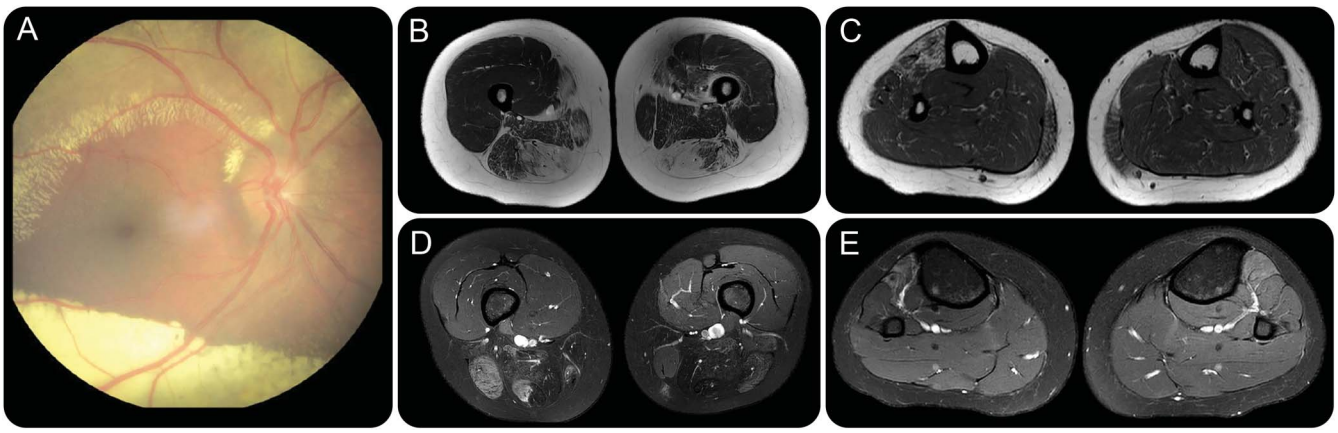
Figure Funduscopic evaluation and muscle MRI studies in facioscapulohumeral muscular dystrophy



(A) Funduscopic analysis showing retinal atrophy and massive exudate deposition in peripheral retina. Axial muscle MRI disclosing fatty replacement in posterior thigh muscles (semimembranous and semitendinosus) and in right anterior tibial compartment in T1-weighted sequences (B, C) and T2-weighted sequences with fat suppression (D, E).

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Neurology 2016;87;e39
DOI 10.1212/WNL.0000000000002899